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Short Report

Sudden death due to aortic rupture complicating undiagnosed coarctation of the aorta in a teenager – A case report and review of the literature

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Abstract

Coarctation of the aorta is one of the more common congenital cardiac defects accounting for between 5 and 10% of cases of congenital heart disease. It has traditionally been divided into infantile (pre-ductal) and adult (ductal) types. Prior to the development of surgical treatment for coarctation, the condition was associated with significant morbidity and mortality with the most common causes of death being aortic rupture, congestive cardiac failure, endocarditis and intracerebral haemorrhage. Presentation of undiagnosed aortic coarctation as sudden and unexpected death is today a distinctly uncommon phenomenon. We report one such case, that of an adolescent male who at autopsy was noted to have coarctation of ductal type with aortic dissection and haemopericardium. The presence of this condition should be considered by the forensic pathologist confronted with aortic dissection, particularly in a young person.

Keywords: Aorta; Coarctation; Sudden; Death; Teenager

1. Introduction

Coarctation of the aorta is an uncommon condition which accounts for between 5% and 10% of all cases of congenital heart disease. Cases are traditionally classified into infantile (preductal) or adult (ductal) based on the site of obstruction.[1] Since the time of publication of the first typical case of adult type coarctation in 1791 by Paris,[2] the natural history of the condition has been well documented. In fact in 1930, Abbott in her seminal review observed that 'few subjects in the literature have been treated so thoroughly from the bibliographical standpoint[2] Notwithstanding this, since the advent of surgical treatment in 1945,[3] reported cases of sudden unexpected death due to undiagnosed coarctation are rare and those reported have usually occurred in the hospital setting. In fact, we could find no cases of denovo presentation to the forensic pathologist in the medicolegal setting in the English speaking literature.[4] We report one such case.

2. Background

The Victorian Institute of Forensic Medicine is a purpose built facility in Melbourne Australia providing a medicolegal death investigation service to the coroner for the State of Victoria. The latter is charged with the investigation of prescribed categories of so-called reportable deaths of which sudden, unexpected and traumatic deaths form a significant proportion.[5] The case at hand involves a

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17-year-old boy with no past medical history who collapsed and died at home. The investigation of his death fell under the purview of the coroner.

3. Postmortem examination

The deceased was a 17-year-old male of weight 73 kg and height 183 cm. He had no past medical history and in particular he had never been noted to have a heart murmur or to be hypertensive and his exercise tolerance was good with no reported syncopal episodes. External signs of trauma were lacking. Significant findings on internal examination were confined to the cardiovascular system. The heart was enlarged weighing 522 g (expected weight based on body weight 336 g with upper 95% confidence limit of 444 g and based on height 341 g with upper 95% confidence limit of 481 g).[6] The left ventricle showed concentric hypertrophy, but there was no evidence of chamber dilatation. The myocardium showed evidence neither of recent nor remote ischaemic damage. The cardiac valves were normal and in particular the aortic valve, which was tricuspid, showed no evidence of stenosis. Coronary artery anatomy was normal and the vessels free of atheroma.. There was a haemopericardium of 750 mls, which comprised blood and clot. The ascending thoracic aorta was dilated and the serosa markedly haemorrhagic. A 3.5 cm ragged linear defect was noted in the intimal surface and blood had tracked intramurally and proximally to the region of the aortic valve (where it had ruptured into the pericardial sac) and distally to the aortic arch. Within the descending thoracic aorta, in the region of the ligamentum arteriosum, a tight stenotic hourglass like segment was observed and which allowed the passage of a 2 mm diameter probe only with some difficulty (Fig. 1). The aorta proximal to the site of narrowing was dilated and described as 'thin walled and floppy' whilst distal to the obstruction the aorta was macroscopically normal with the exception of dilated posterior intercostal artery ostia. The features were considered typical of coarctation of the aorta of adult or ductal type. The remainder of the internal examination was unremarkable with no aneurysms noted on the cerebral vasculature Notching of ribs posteroinferiorly was not observed although neither antemortem nor postmortem radiographs were available for examination.

Extensive histological examination was undertaken. The myocardium of the left ventricle showed patchy interstitial fibrosis most pronounced in a subendocardial location, but no evidence of acute ischaemic damage. The thoracic aorta proximal to the site of coarctation was markedly abnormal. Dissection of blood was observed within the outer third of the media and prominent pools of acid mucosubstance (highlighted by way of PAS–AB (Periodic acid Schiff–Alcian Blue) stain were noted (Fig. 2). An EVG (Elastic Van Giessen) stain revealed pronounced fragmentation of the elastica. There was no evidence of aortitis. Distal to the site of coarctation the aorta was microscopically normal and the kidneys showed no evidence of hypertensive type changes. Routine toxicological screening for alcohol and common drugs of abuse was negative. Death was ascribed to cardiac tamponade due to haemopericardium complicating dissection of the ascending thoracic aorta in the setting of undiagnosed coarctation of the aorta of adult (or ductal) type.

4. Discussion

Coarctation of the aorta accounts for between 5% and 10% of all congenital heart defects and is an important treatable cause of secondary hypertension. It has historically been divided into infantile (or preductal) and adult (or ductal) types based on the site of obstruction with reference to the ductus arteriosus although Rao argues that a 'detailed review of the anatomy suggests all coarctations are juxtaductal'.[7] The classification is not without its shortcomings as the adult type has been described in infants[8] and the infantile in adults. Moreover, some of the older authorities argue that combined forms can occur.[9] The

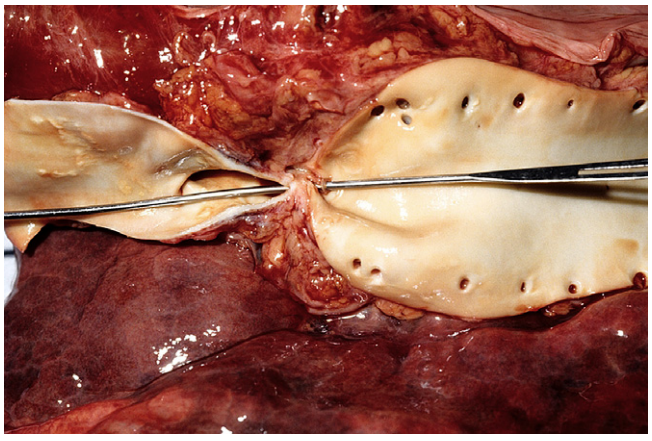

Fig. 1. Macroscopic photo of tight coarctation of descending thoracic aorta with prominent intercostal artery ostia distal to site of narrowing.

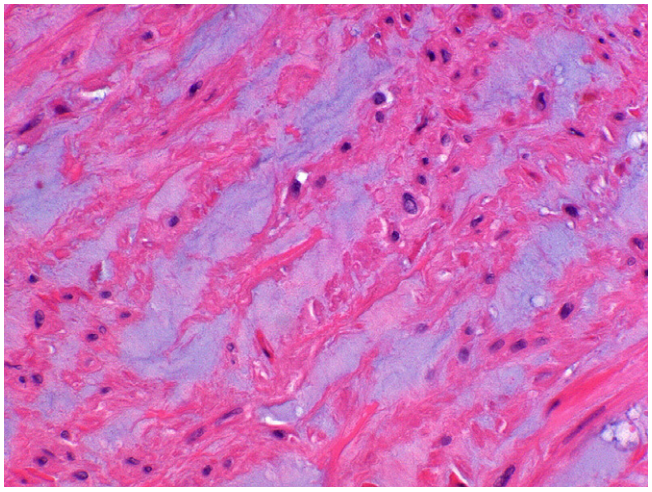

Fig. 2. Aorta proximal to site of coarctation. H and E × 400.

condition is more common in males although this gender imbalance is less pronounced in infants. The infantile type is often associated with significant cardiac defects and the child usually presents in the first year of life in heart failure. The adult type on the other hand tends to be less severe and in up to 20% cases the diagnosis is first made in children and young adults. Other conditions that are well described as occurring in association with coarctation of the aorta include Noonan's and Turner's syndromes, bicuspid aortic valve and cerebral aneurysms.[10] With respect to pathogenesis, there is some uncertainty with haemodynamic and ectopic ductal tissue theories variously invoked.[11]

4.1. Presentation and assessment

We intend to confine our discussion to the adult or ductal type of this disorder. The typical presentation in children, adolescents or young adults is with hypertension and a systolic murmur. Characteristically there may be upper limb hypertension whilst blood pressure measured in the legs is lower. Radiofemoral delay is a classical sign. In addition there may be left ventricular heave or an ejection systolic murmur, the latter usually best heard in the aortic area, but sometimes also in the left interscapular region. The electrocardiograph may show changes in keeping with left ventricular hypertrophy whilst a chest X-ray might reveal cardiomegaly or the characteristic rib notching due to the expansion of collateral circulation (to allow blood from the upper body to reach the lower body by bypassing the obstruction). The latter finding, representing erosion of the inferior aspect of the ribs posteriorly secondary to enlargement of the posterior intercostal arteries, has even allowed the presumptive diagnosis of aortic coarctation to be made by anthropologists examining skeletal remains, a point of some forensic significance.[12] The importance of the role played by primary care physician or general practitioner in early diagnosis of this eminently treatable condition should not be understated.[13] More sophisticated investigations might include echocardiography, angiography or cardiovascular magnetic resonance imaging (CMR) the latter regarded as the imaging procedure of choice notwithstanding its limited availability and possible contraindications in some patients.[14]

5. Natural history

The introduction in 1945 of surgical treatment for coarctation, by Craaford and Nylin,[3] dramatically changed the outlook for patients who had previously succumbed to a variety of complications of their untreated condition, often by early adulthood. In the first half of the twentieth century there appeared two classic papers reviewing the results of postmortem examinations in patients with aortic coarctation. Abbott[2] in her seminal work of 1928 documented the results of some 200 necropsies and provided a comprehensive review of reported cases. Reifenstein[9] and colleagues collected in 1947 the results of an additional 104 autopsies. In an important paper, Campbell[15] summarises and analyses the results of those 304 autopsies on patients with coarctation comprising 'practically all that had been reported up to the time when the introduction of operations made a study of its natural history much more difficult'. The most common causes of death in descending order of frequency were congestive cardiac failure, aortic rupture, bacterial endocarditis and intracranial haemorrhage with almost half the patients succumbing before the age of thirty (Table 1).

Given the development of increasingly sophisticated treatment options for coarctation and the ever dwindling hospital (or non medicolegal) autopsy rate there is little doubt that the described autopsy series are unlikely to be surpassed in terms of providing an understanding of the pathology and natural history of the condition in its undiagnosed and untreated form.

6. Treatment

Descriptions of surgical treatment for coarctation of the aorta first appeared in the literature in the mid 1940s[3,16–18] and the procedures are even today not without risk. Major complications include intra-operative death, paraplegia, recurrent or residual coarctation and aneurysm formation. Late complications can also occur such as premature coronary artery disease, cerebrovascular accident and aortic dissection. There are other treatment options available including balloon angioplasty[19] and endovascular stenting.[20] The optimal treatment option for an individual patient will depend on a variety of factors including age, coarctation morphology, whether previous surgery has occurred and the results of the institution with different treatments.[21]

There have been various case reports of sudden unexpected death in patients who had been previously diagnosed and treated for coarctation. Aortic dissection following patch aortoplasty[22] and coartectomy[23] have both been described. In one of the more extensive reviews of morbidity and mortality from aortic rupture in children with coarctation, Nikaidoh and colleagues[24] reviewed some 325 patients treated at Children's Memorial Hospital in Chicago over a 29-year period. They identified three cases of aortic rupture, two associated with mycotic aneurysm

Table 1
Causes of death in coarctation of aorta

Cause of death	Number	Percentage	Mean age	Usual decade
Congestive cardiac failure	78	26	39	3rd–5th
Incidental to coarctation	80	27	47	4th–6th
Aortic rupture	64	22	25	2nd and 3rd
Bacterial endocarditis or aortitis	37	12.5	29	First five
Intracranial haemorrhage	37	12.5	29	2nd and 3rd

Distilled from works of Abbott,[2] Reifenstein[9] and Campbell.[15].

and one with aortic dissection. There was a fatal outcome in two of the children. Of note the diagnosis of coarctation had been made at birth in the child who ultimately succumbed to a dissecting aneurysm. The authors note that patients with undiagnosed or neglected coarctation are not surprisingly at risk of the same complications that would befall patients in the era preceding refinement of surgical treatment options.

7. Conclusion

Sudden unexpected death in children and young adults due to undiagnosed natural processes is extremely uncommon and can prove a diagnostic challenge to the practicing forensic pathologist. When confronted with unsuspected aortic dissection in the child, teenager or young adult and in the absence of major trauma, the pathologist will often turn his or her mind to the possible existence of a disorder of connective tissue such as Marfan syndrome or one of the variants of Ehlers–Danlos syndrome and the significant attendant ethicolegal and genetic implications of such a diagnosis for surviving family members.[25] But aortic dissection is also a well described complication of coarctation of the aorta accounting for up to 20% deaths in the days before surgical treatment was an option. It also can develop as a late complication in treated patients. Whilst developmental anomalies of the cardiovascular system in medicolegal autopsy practice are often the purview of the paediatric pathologist, our case is a timely reminder to the forensic pathologist, that when confronted with a case of sudden unexpected death due to dissecting aortic aneurysm, the underlying disease process may in fact represent an extremely uncommon presentation of coarctation of the aorta, one of the more common manifestations of congenital heart disease.

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